San Francisco
September 10, 2004

Large Formal Ontologies for Biomedicine

Panel

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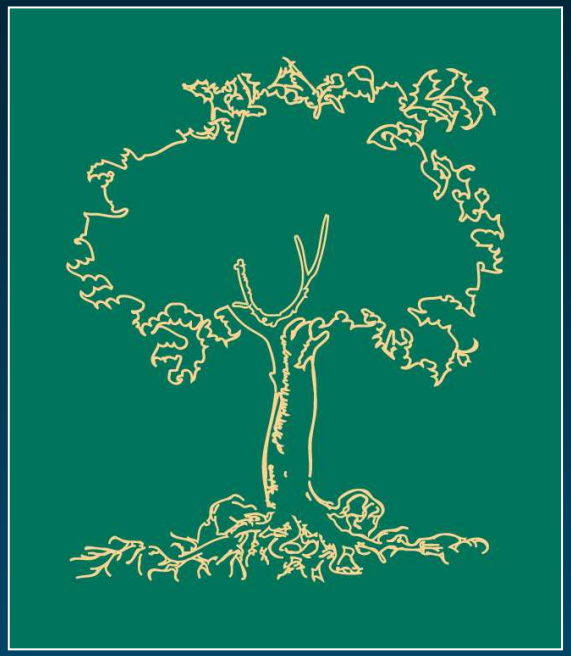
What does UMLS stand for?

- Unified
- Medical
- Language
- System



Motivation

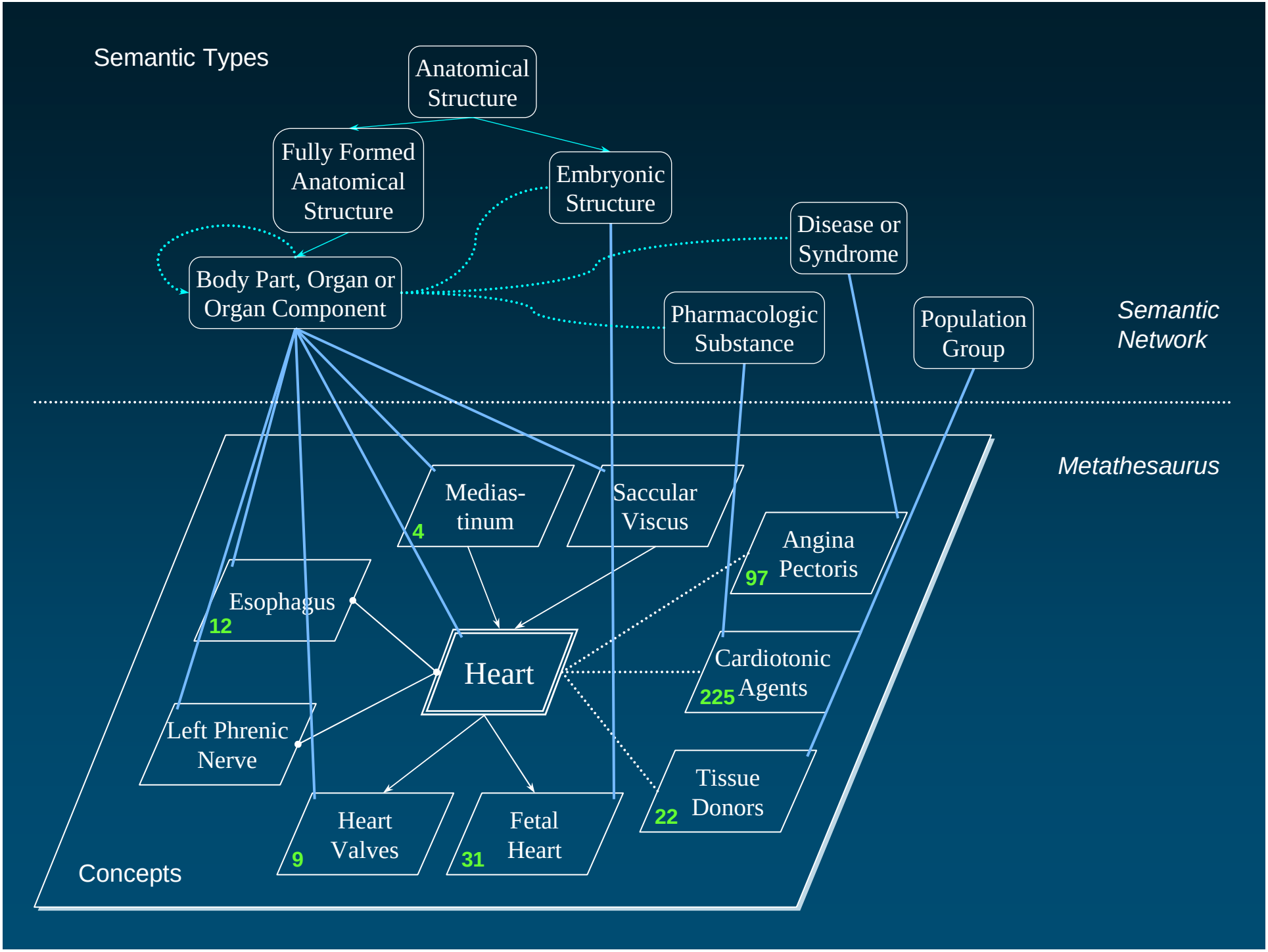
- Started in 1986
- National Library of Medicine
- “Long-term R&D project”

«[…] the UMLS project is an effort to overcome two significant barriers to effective retrieval of machine-readable information.

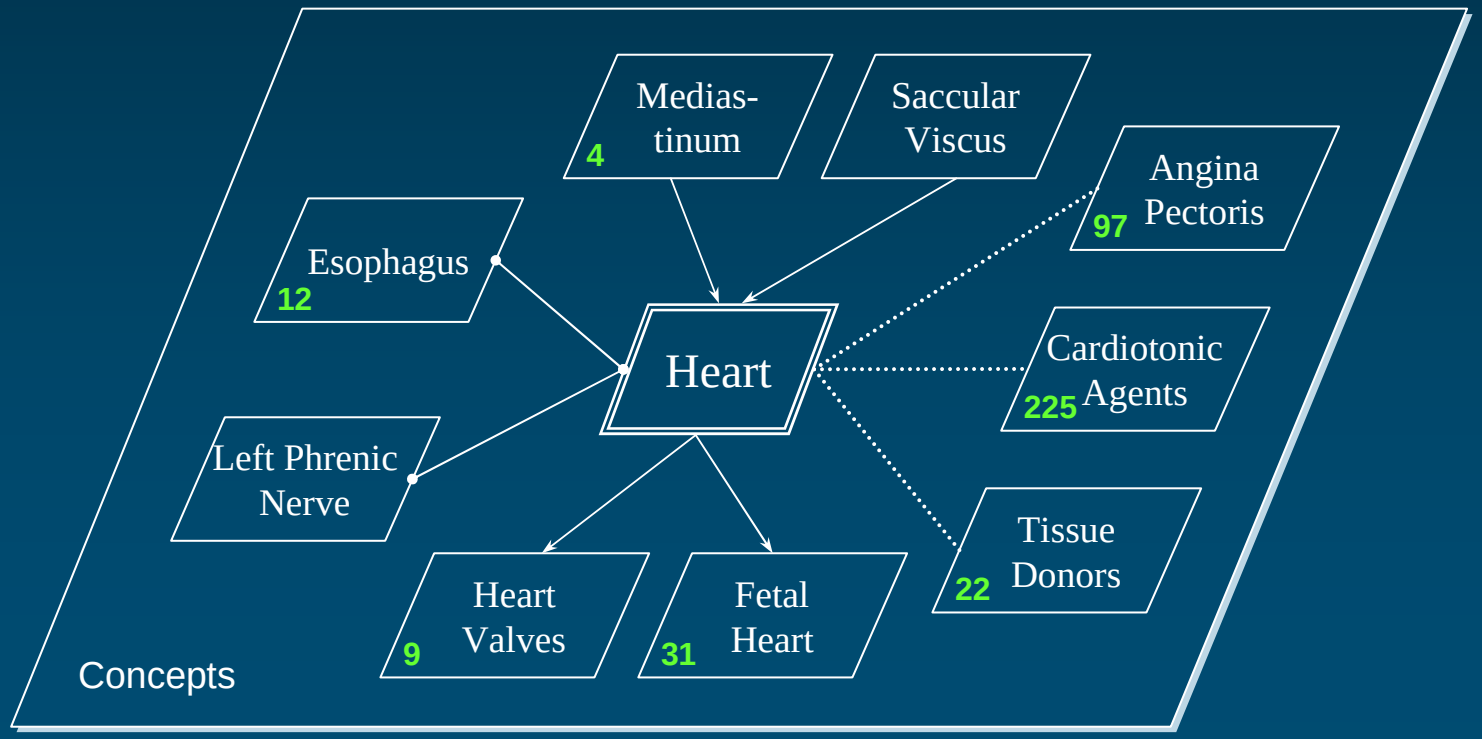
- The first is the variety of ways the same concepts are expressed in different machine-readable sources and by different people.
- The second is the distribution of useful information among many disparate databases and systems.»

UMLS 3 components

- Metathesaurus
  - Concepts
  - Inter-concept relationships
- Semantic Network
  - Semantic types
  - Semantic network relationships
- Lexical resources
  - SPECIALIST Lexicon
  - Lexical tools


Semantic Types
Anatomical Structure
Fully Formed Anatomical Structure
Embryonic Structure
Disease or Syndrome
Body Part, Organ or Organ Component
Pharmacologic Substance
Population Group
Semantic Network
Metathesaurus
Medias-tinum
4
Saccular Viscus
Angina Pectoris
97
Esophagus
12
Heart
Cardiotonic Agents
225
Left Phrenic Nerve
Tissue Donors
22
Heart Valves
9
Fetal Heart
31
Concepts

UMLS Metathesaurus

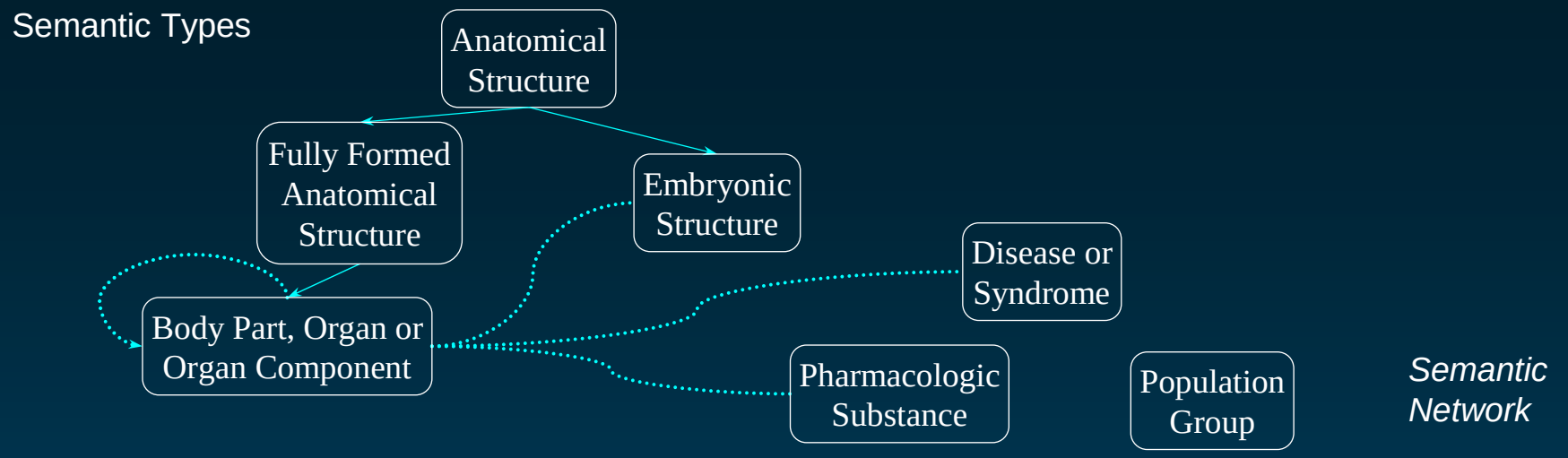


Metathesaurus Large (2004AB)

- 134 source vocabularies
  - 73 families of vocabularies
- 1M concepts
  - 4.3M distinct concept names
- 15M relations

Metathesaurus Essentially informal

- By design
  - Terminology integration project
- By mandate
  - Vehicle for what is asserted in the source vocabularies
- Because if its purpose
  - Influenced by information retrieval
- No formalism
- Manual curation, but insufficient guidelines



UMLS Semantic Network

Semantic Network More formal

- Manually created
  - Regardless of the position of the corresponding terms in existing vocabularies
- No particular formalism
  - Triples <ST_1, *rel*, ST_2>
- Quantification: some value from
  - <*Drug*, *treats*, *Disease or Syndrome* >
  - some *Drug treats* some *Disease or Syndrome*

Semantic Network But small

- 135 semantic types
- 54 relationships
- 558 relations

Can the Metathesaurus be more formal?

- Probably not given its design
- Trade-offs
  - Ingredient/drug synonymy
  - Clinical synonymy/linguistic synonymy
- But some recent changes in its representation may help
  - Source transparency: relations are no longer recorded at the concept level, but at the atom level

Couldn't the Metathesaurus use another formalism (e.g., OWL)?

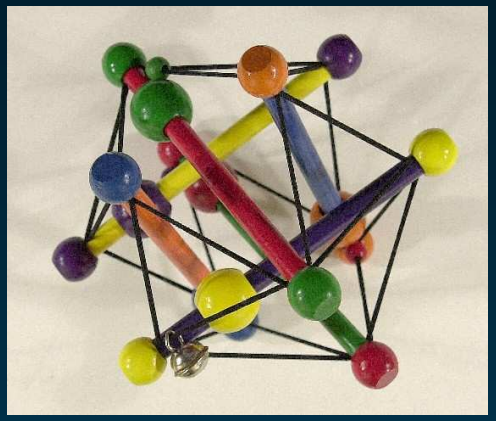
- Simply changing the formalism would not help
- What is needed are more precise, better defined relationships (isa vs. parent/child)
  - A trivial change of formalism would (wrongly) assume isa for each parent/child relationship
- Transforming portions of the UMLS in DL is resource intensive
- The OWLization of other terminology systems has not produced convincing results

Towards more consistency

- Many studies have investigated consistency issues in the Metathesaurus
- Some have developed algorithms, e.g.,
  - Identify and remove circular hierarchical relations
  - Use Semantic Network relations to assess the validity of Metathesaurus relations
  - …

Conclusions

- Metathesaurus
  - Large and informal
  - But could be used as the basis for deriving an ontology of biomedicine
    - Manual curation
    - Lexical methods for acquiring ontological relations from terminlogy
    - Would still require an upper-level ontology
    - And a formalism



Medical Ontology Research

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